THE IMPROVED DIASORIN Q-LAMP ASSAY FOR THE ACCURATE AND ULTRA-FAST DETECTION OF COMMON AND RARE ISOFORMS OF THE BCR-ABL1 TRANSLOCATION

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The molecular detection of BCR-ABL1 transcripts by RT-PCR is mandatory for the diagnosis of Ph+ Leukemias at onset. Recently a faster and reliable assay based on the Q-LAMP technology developed by DiaSorin has entered in laboratory routine. This assay detects in one hour BCR-ABL1 p190 (e1a2) and p210 (e13a2, e14a2) isoforms. In this study, we evaluated the new improved Q-LAMP formulation designed to detect also less frequent isoforms of the BCR-ABL1 transcripts p190 and p210 (e1a3, e13a3, e14a3). In addition, clinical studies demonstrated that the assay is capable of detecting also the rare isoform p230 (e19a2, e19a3). Methods: The new Q-LAMP technology consists in a multiplex assay for the differential detection of p190 and p210 transcripts and the amplification of the GUSB endogenous RNA. The assay has been tested on 185 clinical samples including 95 p210 positive (57 e13a2 and 28 e14a2, 8 e13a3 and 2 e14a3), 38 p190 positive (33 e1a2 and 5 e1a3) and 50 BCR-ABL1 negative samples. Additional 2 p230 rare isoforms were also included in this study. All samples were previously tested by RT-PCR, considered as the reference method. Results: The new BCR-ABL Q-LAMP assay showed 100% concordance with the RT-PCR, with an expected delayed amplification time for rare isoforms respect to the common ones. The average amplification time of p210 common isoforms were 22,24 and 25,03 min compared to the p210 and p190 rare isoforms that showed 26,54 and 36,84 min, respectively. The 2 p230 (e19a2) rare isoforms were also tested and resulted valid although, due to the very long transcript, they showed a very high average amplification time (50 and 48 min). Moreover, we observed an interesting discrimination between the e13a2 and the e14a2 isoforms in terms of amplification times (20,21 versus 26,36 min) likely associated to the different length of the two transcripts, with low coefficients of variability (0,15 and 0,11 respectively). Conclusions: The enhanced BCR-ABL Q-LAMP assay well proved to detect both common and uncommon isoforms of the BCR-ABL1 translocation. This improved performances, combined with the speed and the close tube format, allow laboratories to optimize their workflow and represent a reliable solution for molecular diagnosis of Philadelphia Positive Leukemias.